FOXP3 gene

forkhead box P3

Normal Function

The *FOXP3* gene provides instructions for producing the forkhead box P3 (FOXP3) protein. The FOXP3 protein attaches (binds) to specific regions of DNA and helps control the activity of genes that are involved in regulating the immune system. On the basis of this role, the FOXP3 protein is called a transcription factor. This protein is essential for the production and normal function of certain immune cells called regulatory T cells, which play an important role in preventing autoimmunity. Autoimmunity occurs when the body attacks its own tissues and organs by mistake. The FOXP3 protein is found primarily in an immune system gland called the thymus, where regulatory T cells are produced.

Health Conditions Related to Genetic Changes

immune dysregulation, polyendocrinopathy, enteropathy, X-linked syndrome

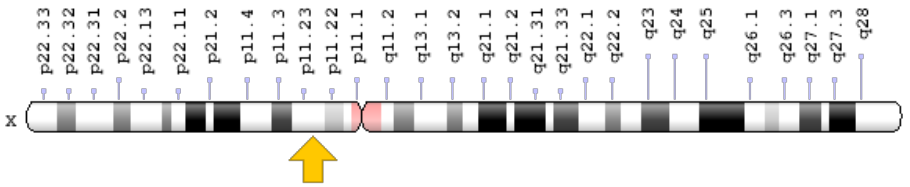
At least 21 mutations in the *FOXP3* gene have been found to cause immune dysregulation, polyendocrinopathy, enteropathy, X-linked (IPEX) syndrome. Most mutations change one protein building block (amino acid) in the region of the FOXP3 protein that binds to DNA or lead to the production of an abnormally short, nonfunctional protein. Mutations in the *FOXP3* gene result in reduced numbers or a complete absence of regulatory T cells. Without the proper number of regulatory T cells, the body cannot control immune responses. Normal body tissues and organs are attacked, causing the multiple autoimmune disorders present in people with IPEX syndrome.

type 1 diabetes

Chromosomal Location

Cytogenetic Location: Xp11.23, which is the short (p) arm of the X chromosome at position 11.23

Molecular Location: base pairs 49,250,436 to 49,266,505 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AIID
- DIETER
- FOXP3_HUMAN
- immune dysregulation, polyendocrinopathy, enteropathy, X-linked
- immunodeficiency, polyendocrinopathy, enteropathy, X-linked
- IPEX
- JM2
- MGC141961
- MGC141963
- PIDX
- scurfin
- XPID

Additional Information & Resources

Educational Resources

- Immunobiology (fifth edition, 2001): Autoimmune Responses are Directed Against Self Antigens
  https://www.ncbi.nlm.nih.gov/books/NBK27155/

GeneReviews

- IPEX Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1118

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28FOXP3%5BTIAB%5D%29+OR+%28forkhead+box+P3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D

OMIM

- FORKHEAD BOX P3
  http://omim.org/entry/300292

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/FOXP3ID44129chXp11.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=FOXP3%5Bgene%5D
- HGNC Gene Family: Forkhead boxes
  http://www.genenames.org/cgi-bin/genefamilies/set/508
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=6106
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/50943
- UniProt
  http://www.uniprot.org/uniprot/Q9BZS1

Sources for This Summary

- Fuchizawa T, Adachi Y, Ito Y, Higashiyama H, Kanegane H, Futatani T, Kobayashi I, Kamachi Y, Sakamoto T, Tsuge I, Tanaka H, Banham AH, Ochs HD, Miyawaki T. Developmental changes of FOXP3-expressing CD4+CD25+ regulatory T cells and their impairment in patients with FOXP3 gene mutations. Clin Immunol. 2007 Dec;125(3):237-46. Epub 2007 Oct 3.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17916446
- GeneReview: IPEX Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1118

- Nik Tavakoli N, Hambly BD, Sullivan DR, Bao S. Forkhead box protein 3: essential immune regulatory role. Int J Biochem Cell Biol. 2008;40(11):2369-73. Epub 2007 Oct 10. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18037337
- Torgerson TR, Ochs HD. Immune dysregulation, polyendocrinopathy, enteropathy, X-linked: forkhead box protein 3 mutations and lack of regulatory T cells. J Allergy Clin Immunol. 2007 Oct; 120(4):744-50; quiz 751-2. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17931557

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/FOXP3

Reviewed: March 2013
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services